

ABSTRACT

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Title: Current possibilities of laboratory diagnostics of anemia

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This bachelor's thesis discusses the general informations and summary of etiopathogenetical classification of anaemia and possibilities of laboratory diagnostics. Thesis is mainly focused on laboratory diagnosis of inherited types of anaemia.

Anaemia or anaemia syndrome is a pathological condition characterized by low levels of haemoglobin and hematocrit under the physiological limit. Clinical manifestations are variously severe depending on development of disease and type of anaemia. Anaemia patient suffer from reduced oxygenation of tissues which determinates many symptoms of tissue hypoxia. Other symptoms are associated with hypovolemia and hyperkinetic circulation that can occur. In haemolytic anaemia, patients have symptoms of intravascular or extravascular haemolysis.

Primary (hereditary) anaemia are congenital, the cause lies in genetic mutations that result a changes of phenotype properties of erythrocytes.

The process of diagnosis determinating includes correct identification of family history, relevant medical examinations and laboratory testing methods, which are essential part of modern medicine. Important are methods such as routine morphological examinations (blood counts, blood cell morphology) or specific laboratory tests (cytogenetic and molecular genetic methods), biochemical parameters examination and microscopic examination of bone marrow. All the examinations have great importance in the diagnosis.

Providing of genetic counselling have a importance role. It includes education and counselling on issues of inherited disease and psychosocial support. It is available at the department of clinical and medical genetics in health care facilities.

Correct and early diagnosis contributes to a better prognosis due to early initiation of treatment options. In the case of genetically inherited anaemia, there is available replacement therapy, the most common is the administration of transfusion blood products.